MS/JAMA — **Essay**
Vol. 280, pp. 1533-1534, Nov 4, 1998

Genetics in the Context of Medical Practice

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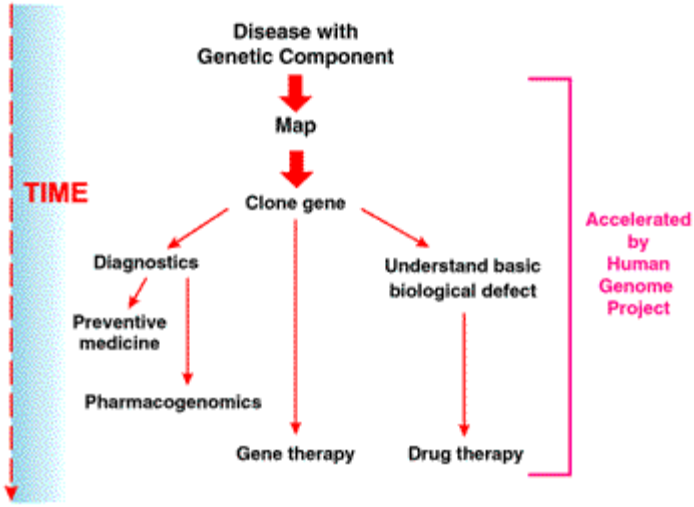
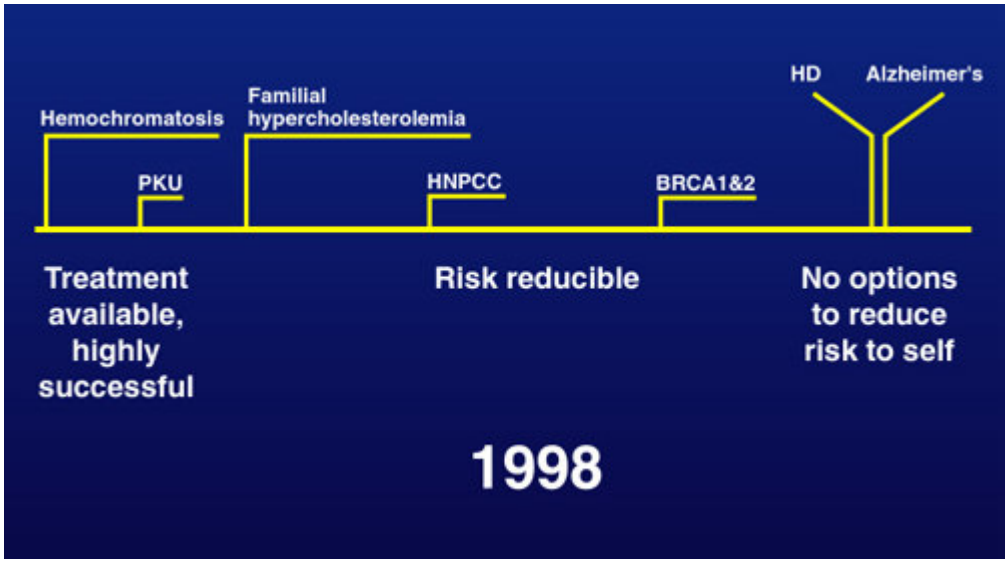
The Human Genome Project (HGP) is an international scientific effort to map and sequence the entire human genome. Since its inception in the United States in 1990, as a joint effort by the National Institutes of Health (NIH) and Department of Energy (DOE), the HGP now includes contributions from genome centers in the United Kingdom, France, Canada, Germany, and Japan. In September 1998, the NIH and DOE announced an accelerated timetable for sequencing the genome, and the entire human sequence is expected to be completed by the end of 2003. This information will benefit clinical medicine by enabling physicians to diagnose and treat heritable disorders more effectively.



Courtesy of Jane Ades, NIH

From Maps to Medicine

Information from the HGP has accelerated the rate of gene discovery. Once a disease gene is identified, DNA-based diagnostic tests can be developed to detect at-risk individuals. Knowledge of a patient's genetic makeup can allow physicians to minimize disease risk through preventive medicine and conventional drug therapies. A more novel treatment is gene therapy, which compensates for the defective gene by providing an exogenous functional copy. Another promising tool is pharmacogenomics, where a person's genotype is used to predict those pharmaceuticals that will prove most therapeutic and identify those that could be deleterious. For example, the cholinesterase inhibitor tacrine appears to be less effective in Alzheimer disease patients who carry the apolipoprotein E4 allele.



Courtesy of Jane Ades, NIH

Defining the Role of the Primary Care Physician

As discoveries from the HGP are translated into meaningful medical diagnostics and therapeutics, genetics will heavily influence clinical decision making. As the number of treatable genetic diseases increases, physicians will need to use and interpret genetic tests correctly, determine those genetic treatments that are available, and learn how to access these services. Perhaps the most important role for the primary care physician is first to identify a potential genetic disorder. Hence, physicians must be prepared to integrate information derived from a careful family history with the molecular data provided by the HGP.

Genetics Education for Physicians

A recent American Medical Association (AMA) survey indicated that 59% of Americans are somewhat or very likely to take advantage of genetic testing and that 72% believe that their primary care physician can interpret these results. However, in a recent study, physicians misinterpreted nearly one third of predictive test results for colon cancer, and fewer than 20% of patients received appropriate genetic counseling.

To strengthen genetics knowledge among physicians, recent guidelines by the American Society of Human Genetics have concentrated on increasing the emphasis on genetics in medical school curricula. To ensure quality continuing genetics education for health care professionals, the National Coalition for Health Professional Education in Genetics was developed in 1996 by the AMA, the American Nurses Association, and the NHGRI to provide genetics information online, better represent genetics on licensing examinations, and facilitate the development of core curricula in genetics.

On the Safe Use of Genetic Tests

In 1997, the NIH-DOE Task Force on Genetic Testing issued a set of recommendations to ensure the safety and proper use of genetic tests prior to their use in a clinical setting. Recommendations include to (1) establish an Advisory Committee on Genetic Testing in the Office of the Secretary of Health and Human Services (HHS); (2) establish a means for prioritizing genetic tests in high- and low-scrutiny categories; (3) require that diagnostic labs setting up a genetic test design an institutional review board–approved protocol for collecting data on analytic and clinical validity;

(4) recommend external review of protocol outcomes prior to marketing; (5) emphasize the need for public and professional education; and (6) emphasize the need for special consideration for testing rare diseases. Based on this report, the federal Advisory Committee on Genetic Testing, which will report to the HHS Secretary, was chartered in August 1998.

Protecting Patient Rights

A recent survey indicated that nearly 7 out of 10 Americans are somewhat or very concerned that genetic information may be used against them by either their employer or health insurance provider.[6] In 1995, a set of recommendations to lawmakers dealing with issues pertaining to health insurance and genetic discrimination was compiled by the NIH-DOE Working Group on Ethical, Legal and Social Implications of Human Genome Research and the National Action Plan on Breast Cancer. These guidelines would prohibit insurance providers from increasing premiums or determining eligibility based on predictive genetic information and would prohibit insurance providers from accessing or disclosing genetic information. A major step in this direction came in 1996 with passage of the Health Insurance Portabil